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Case Report Open Access

Comparing Structure-Function Correlations in Superior Segmental Optic Nerve Hypoplasia and Juvenile Open Angle Glaucoma Using Spectral-Domain Optical Coherence Tomography

Sirinya Suwannaraj, Kara M. Cavuoto* and Ta C. Chang

Bascom Palmer Eye Institute, University of Miami Miller School of Medicine, Department of Ophthalmology, Miami, Florida, USA

***Corresponding author:** Kara M. Cavuoto, M.D., Bascom Palmer Eye Institute, University of Miami Miller School of Medicine, Department of Ophthalmology, Miami, Florida, 900 NW 17th St, Miami, Florida, 33136, USA, Tel: 305-326-6324; Fax: 305-547-3675; E-mail: kcavuoto@med.miami.edu

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Abstract

Superior segmental optic nerve hypoplasia (SSOH) is a congenital anomaly of the optic nerve that is commonly misdiagnosed as normal tension glaucoma (NTG) and/or juvenile open angle glaucoma (JOAG). We demonstrate the utility of SD-OCT in assessing the structure-function correlation when differentiating between SSOH and JOAG.

Keywords Superior segmental optic nerve hypoplasia; Glaucoma; Imaging; Retinal nerve fiber layer optical coherence tomography

Case Report

Case 1

A 13 year-old male with a history of an increased cup-to-disc ratio of the left eye was referred for evaluation of JOAG. He had no family history of glaucoma. On examination, his visual acuity was 20/25 in both eyes. Applanation tonometry revealed an intraocular pressure (IOP) of 14 mmHg on the right without treatment and 12 mmHg on the left with topical beta-blocker treatment. Records of the IOP prior to presentation and treatment were unavailable.

Gonioscopy showed open angles in both eyes. Fundus examination revealed a normal optic disc with a superior peripapillary scleral halo in the right eye and superior disc pallor with thinning of the superior rim in the left eye (Figure 1a). A Humphrey visual field 24-2 (Carl Zeiss Meditec, Dublin, CA) showed an inferior field defect in left eye (Figure 1b), while SD-OCT showed RNFL thinning superiorly (Figure 1c). The patient was diagnosed with SSOH of the left eye and was instructed to discontinue the topical beta-blocker. In twenty months of follow up, the untreated IOP has remained within physiologic range and serial visual fields and SD-OCT RNFL analyses have remained stable on two subsequent tests.

Case 2

A 13 year-old female was referred for evaluation after a failed vision screening. She was asymptomatic and had no personal or family history of glaucoma. On examination, her best corrected visual acuity was 20/30 in both eyes. Her IOP by applanation was 19 mmHg in both eyes. Pachymetry showed a central corneal thickness of 640 μm in the right eye and 632 μm in the left eye. Gonioscopy showed open angles to ciliary body without abnormal iris insertions and no stigmata of anterior segment dysgenesis in both eyes.

Fundus examination revealed optic disc cupping of 0.6 in the right eye and 0.4 in the left eye, with bilateral thin superior rims (Figure 2a). A Humphrey visual field 24-2 showed bilateral inferior visual field defects (Figure 2b). SD-OCT showed bilateral thinning of superior RNFL in both eyes (Figure 2c). The patient was diagnosed as having bilateral SSOH. No treatment was initiated and the patient has remained stable without visual field or RNFL changes after five months of follow up.

Case 3

An 18 year-old female was referred for urgent glaucoma evaluation. Nine months earlier, she was seen for a second opinion, at which time her IOP by applanation was in the teens on three topical agents without evidence of optic nerve damage (Figure 3a top). On follow up, her visual acuity was 20/20 in both eyes; IOP by applanation was 42 mmHg in the right eye and 40 mmHg in the left eye on maximum topical medications and oral acetazolamide. Gonioscopy showed open angles to ciliary body without abnormal iris insertions and no stigmata of anterior segment dysgenesis in both eyes. Fundus examination revealed optic disc cupping of 0.9 in the right eye and 0.55 in the left eye (Figure 3a bottom).

A follow-up visual field showed dense superior and inferior arcuate defects in the right eye and an early superior visual field defect in the left eye (Figure 3b). SD-OCT showed generalized thinning bilaterally with superimposed superior and inferior RNFL defects in right eye and inferior defect in the left eye (Figure 3c right), markedly worsened since her initial examination nine months prior (Figure 3c left). Based on the constellation of progressive optic nerve cupping, marked ocular hypertension, new visual field defects and RNFL defects corresponding to the visual field defects, the patient was diagnosed with JOAG in both eyes and subsequently underwent circumferential trabeculotomy bilaterally. At two months after surgery, the IOP in both eyes was controlled without the use of medications.

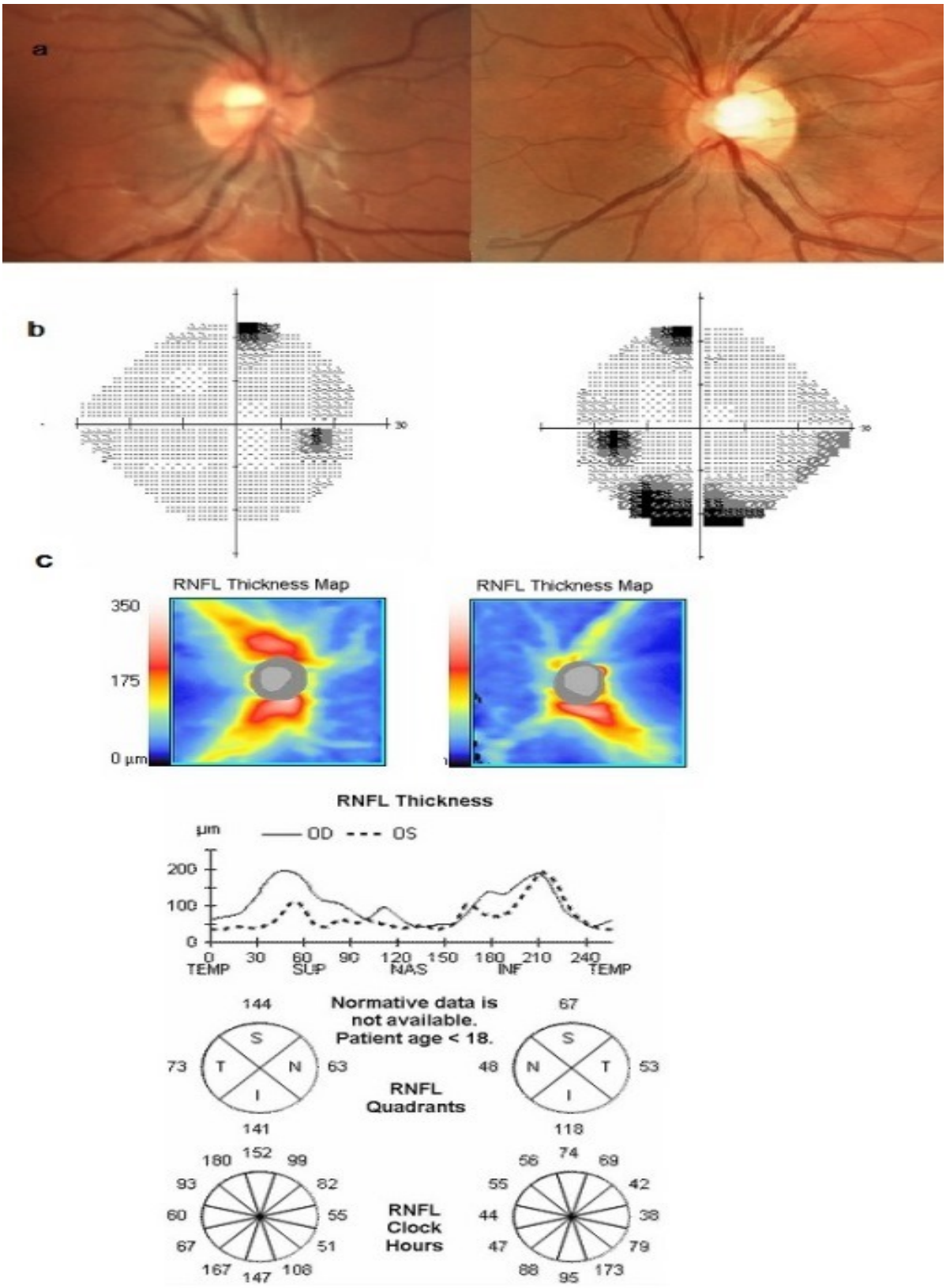

Figure 1: Optic nerve photos **(a)** visual fields **(b)** and optical coherence tomography retinal nerve fiber layer analysis **(c)** of a 13 year-old male with unilateral superior segmental optic nerve hypoplasia of the left eye.

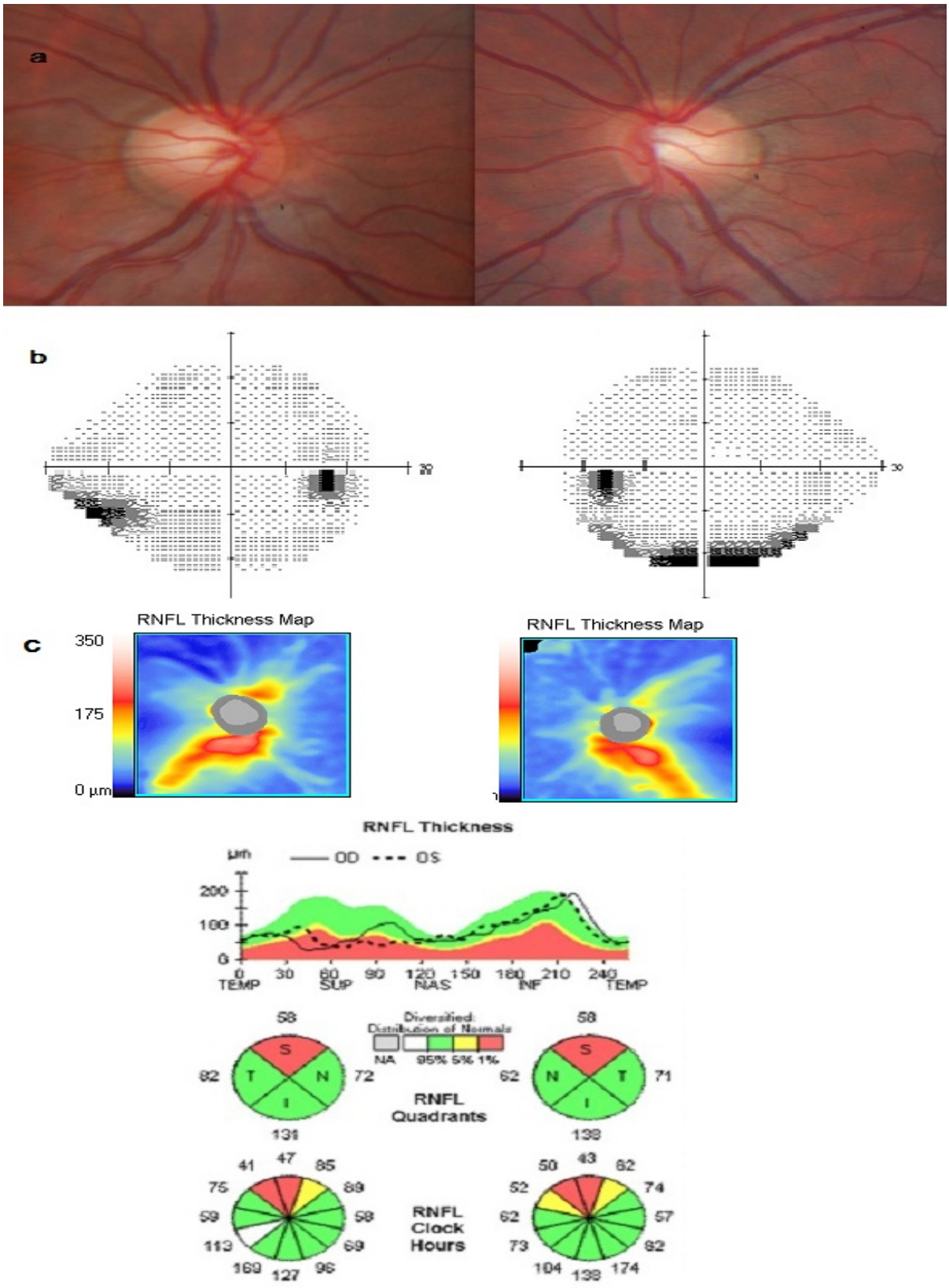

Figure 2: Optic nerve photos **(a)** visual fields **(b)** and optical coherence tomography retinal nerve fiber layer analysis **(c)** of a 13 year-old female with superior segmental optic nerve hypoplasia in both eyes.

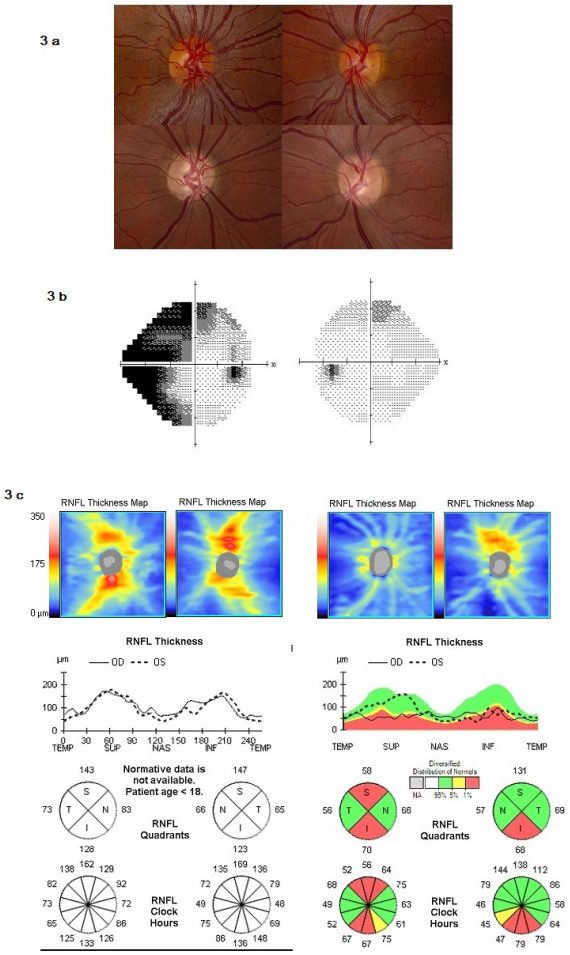

Figure 3: Optic nerve structural and functional studies of 18 year-old female with juvenile open angle glaucoma demonstrating glaucomatous progression–optic nerves photos on initial presentation (**a**, top) and on follow up (**a**, bottom); visual field on follow up (**b**); optical coherence tomography retinal nerve fiber layer analyses on initial presentation (**c**, left) and on follow up (**c,** right).

Discussion

Gardner and Irvine described the first cases of SSOH as a variant of optic nerve hypoplasia with good visual acuity and inferior altitudinal visual field defect in 1977 [1]. The typical disc morphology of SSOH may incorrectly lead to a diagnosis of NTG or JOAG, especially if the IOP is considered borderline. Takagi et al. proposed several key differences between SSOH from NTG: in SSOH, visual fields show no Bjerrum scotoma, disc cupping is not classically glaucomatous and can be out of proportional to the severity of the visual field defect [2]. RNFL defects in SSOH have a sectored pattern, in contrast to the localized slit-like pattern found in NTG [3]. Imaging indices developed for detecting glaucoma will also identify approximately half of the optic discs with SSOH, but the affected sectors may differ. Whereas a notch in the inferotemporal or superotemporal rim is commonly found in glaucoma, it is more commonly noted in the superonasal segment in SSOH, creating a "single hump" pattern [4-5], as seen in cases 1 and 2. Also, the optic nerve head rim and RNFL thickness in children with glaucoma has been found to be reduced concentrically, with particular losses in the superior and inferior quadrants, which would also differ significantly from SSOH [6]. As demonstrated in our series, SD-OCT RNFL analysis provides excellent quantitative measurements of neuroretinal rim volume and RNFL thickness, which correlate well to visual field defects in SSOH. However, characteristics of a single analysis may not differentiate SSOH from JOAG. In case 3, while the SD-OCT RNFL analysis demonstrated generalized thinning in addition to localized RNFL defects, the left eye demonstrate a "single-hump" pattern, albeit in the inferior quadrant. Ultimately, the lack of progression on sequential SD-OCT RNFL analysis and on visual field testing with normal IOP without glaucoma therapy is essential for the diagnosis of SSOH [4].

In summary, when a pediatric patient presents with superior rim thinning with localized loss of superior retinal nerve fiber layer, an inferior visual field defect, and normal IOP, the patient may be monitored closely prior to initiating presumptive IOP-lowering treatment. While SSOH is nonprogressive in nature and is an important entity in the differential diagnosis of childhood glaucoma, there is an isolated case report of SSOH accompanied by NTG in an adult after five years of annual follow up, necessitating long-term monitoring even in low-risk patients [7]. Sequential SD-OCT surveillance and visual field testing are useful tools in detecting progressive glaucomatous changes.

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The authors have no financial disclosures.

Conflict of Interest Statement

The authors have no conflicts of interest.

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